



## BCKDHB gene

branched chain keto acid dehydrogenase E1 subunit beta

### Normal Function

The *BCKDHB* gene provides instructions for making one part, the beta subunit, of an enzyme complex (a group of enzymes that work together). This complex is called branched-chain alpha-keto acid dehydrogenase, or BCKD. Two beta subunits connect with two alpha subunits, which are produced from the *BCKDHA* gene, to form a critical part of the enzyme complex called the E1 component.

The BCKD enzyme complex is responsible for one step in the normal breakdown of three protein building blocks (amino acids). These amino acids—leucine, isoleucine, and valine—are obtained from the diet. They are present in many kinds of food, particularly protein-rich foods such as milk, meat, and eggs. The BCKD enzyme complex is active in mitochondria, which are specialized structures inside cells that serve as energy-producing centers. The breakdown of leucine, isoleucine, and valine produces molecules that can be used for energy.

### Health Conditions Related to Genetic Changes

#### maple syrup urine disease

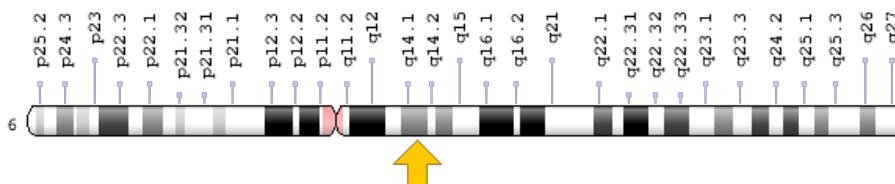
More than 40 mutations in the *BCKDHB* gene have been identified in people with maple syrup urine disease, most often in individuals with the severe, classic form of the disorder. Some of these mutations change single amino acids in the beta subunit of the BCKD enzyme complex. A particular mutation is most common in people of Ashkenazi (eastern and central European) Jewish descent; this mutation replaces the amino acid arginine with the amino acid proline at position 183 in the beta subunit (written as Arg183Pro or R183P). Other *BCKDHB* mutations insert or delete small amounts of DNA in the gene.

Mutations in the *BCKDHB* gene disrupt the normal function of the BCKD enzyme complex, preventing it from effectively breaking down leucine, isoleucine, and valine. As a result, these amino acids and their byproducts build up in the body. This accumulation is toxic to cells and tissues, particularly in the nervous system. The buildup of these substances can lead to seizures, developmental delay, and the other medical problems associated with maple syrup urine disease.

## Chromosomal Location

Cytogenetic Location: 6q14.1, which is the long (q) arm of chromosome 6 at position 14.1

Molecular Location: base pairs 80,106,610 to 80,469,088 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 2-oxoisovalerate dehydrogenase beta subunit
- BCKDH E1-beta
- branched chain keto acid dehydrogenase E1, beta polypeptide
- branched chain keto acid dehydrogenase E1, beta polypeptide (maple syrup urine disease)
- branched chain keto acid dehydrogenase E1, beta polypeptide precursor
- ODBB\_HUMAN

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry (sixth edition, 1998): Major pathways of branched-chain amino acid metabolism (figure)  
<https://www.ncbi.nlm.nih.gov/books/NBK20436/figure/A3097/>
- Basic Neurochemistry (sixth edition, 1998): Maple syrup urine disease was the first congenital defect of branched-chain amino acid catabolism to be described  
<https://www.ncbi.nlm.nih.gov/books/NBK28225/#A3107>

### GeneReviews

- Maple Syrup Urine Disease  
<https://www.ncbi.nlm.nih.gov/books/NBK1319>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28BCKDHB%5BTIAB%5D%29+OR+%28branched+chain+keto+acid+dehydrogenase+AND+E1+AND+beta%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE  
<http://omim.org/entry/248611>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=BCKDHB%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=987](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=987)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/594>
- UniProt  
<http://www.uniprot.org/uniprot/P21953>

## **Sources for This Summary**

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